

[REDACTED]

May 16, 2003

[REDACTED]

RE: [REDACTED]
Date of Birth: [REDACTED]

Dear [REDACTED]

Thank you for referring [REDACTED] for neurologic consultation. As you know, she is a 26-year-old woman who was initially evaluated for leg weakness at Sturdy Memorial by [REDACTED] who found that she had significant proximal bilateral leg weakness. He initiated her nerve conduction studies in the left arm and leg which are essentially normal except for slightly prolonged latencies in the arm. [REDACTED] then asked me if I would continue her study because he thought she might have a myopathy based on the fact that she appeared to have fairly symmetric proximal weakness. I subsequently saw her on May 9, 2003 in the office and did an extensive EMG and found that she had severe chronic neuropathic changes in both lower extremities primarily in the L4-5 muscles but also extending down into the lower myotomes. Based on the findings, additional workup was pursued and she came back today for formal consultation.

She gives a history of having an active sports life in high school where she swam, played crew and ran significantly in college in her freshman year. She did crew and then realized that the commitment was more than she was ready for and stopped doing active sports because of that. She noticed over the following few years that she seemed to be getting weaker in her legs and would work out on machines in attempts to correct that but found that despite working out on weights and such, she gradually had less and less strength in her upper extremities. She denies any history of back pain associated with that or numbness or weakness. She denies bowel or bladder dysfunction. She denies dysfunction in her hands and denies any changes in her bulbar muscles for swallowing and speech.

She has an interesting history of exposure to Lyme disease. By her report, the family lives in Old Lyme, Connecticut and her mother actually had been diagnosed with rheumatoid arthritis and had been on gold injections as well as other cytotoxic and anti-inflammatory meds for several years before she became pregnant although the mother states she was definitely off the gold injections at the time she became pregnant. Retrospectively, in the last six years or so, the mother was diagnosed with chronically high Lyme titers when she saw another physician in Maine where she now lives. Apparently, multiple other family members have been diagnosed with Lyme disease at various points as well. The patient has never had any specific symptoms to support Lyme disease. She has no history of heavy metal exposure although she did drink well water growing up and she does not work around any toxins.

[REDACTED]

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Developmentally, she was noted to have major problems with concentration and also language function that was diagnosed in grade school and she was started on Ritalin with significant improvement in her focusing ability. Her mother has always noticed that when she is not on Ritalin, her ability to carry on a conversation is definitely impaired. There appears to be a speech production issue that responds to Ritalin. The patient has been on Ritalin for over ten years and it has not been recently changed or evaluated. Just recently, she saw a specialist in Boston for reevaluation of her symptoms of ADD.

The mother notes that the patient had obvious clumsiness and difficulty with gross motor skills in kindergarten and first grade so that that is when they first became concerned about her and once she started reading, they realized that she had some sort of learning disability. However, she was able to complete private school and went on to college successfully in spite of it. She now works in a fabric design company.

ALLERGIES: NONE.

EXAMINATION: She is alert and pleasant. She has fluent speech but there appears to be a slight lisp to it which the mother and the patient feel is her baseline speech. There is no nasal quality to it. Attention, concentration and memory appear normal. She is anxious about the tests that are being pursued.

Cranial nerve exam reveals the visual fields are full. The extraocular movements are intact without nystagmus. The pupils are equal and reactive to light and accommodation. The discs are sharp bilaterally with good venous pulsations. There is no facial weakness or numbness. The tongue and uvula are midline. Sternocleidomastoid and trapezius muscles are strong. There are no fasciculations in the tongue. Her hearing is normal.

Motor exam revealed normal tone and full strength in the upper extremities. In the lower extremities, she has 4/5 strength of the hip flexors bilaterally. She does have visible fasciculations in the proximal hip muscles. Otherwise, her strength appeared to be 5/5 throughout. She was able to toe walk and heel walk. However, she could not stand up from a chair without pushing herself with her arms and she could not get off the floor without a modified Gower's maneuver and even with that had some difficulty. Sensory exam is intact to pin, temperature, vibration, and double simultaneous stimulation. She has a negative Romberg. There is no ataxia of finger-to-nose or heel-to-shin. Her gait does reveal slight waddling and she reports some aching of the hips recently with walking.

As noted, her EMG shows remarkable chronic reinnervation in the proximal leg muscles bilaterally worse on the right but also does involve some of the more distal muscles suggesting a multiroot level involvement. The workup she had initially was an MRI of the cervical spine without gadolinium which was normal except for slight levoscoliosis and mild degenerative changes but nothing suggesting cord compression. After seeing me for the EMG, we performed an MRI of the lumbar spine with gadolinium which revealed no enhancing lesions or orthopedic discogenic lesions. She did, however, have a 6 mm extraspinal synovial cyst adjacent to the lateral aspect of the right facet

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joint at L4-5. Blood work initially revealed normal B12 and folate, C-reactive protein, rheumatoid factor and sedimentation rate. Her ANA and Lyme titer are not back as yet and the RPR was negative.

IMPRESSION: This is a most impressive picture of indolently progressive proximal leg weakness with EMG supporting polyradiculopathy in a young woman with a strong history of potential Lyme exposure. I would like to think that this is, in fact, chronic Lyme disease and the patient is scheduled for an LP to address that next week. If the Lyme workup is unrevealing, then she clearly has a strong suggestion of motor neuron disease possible spinal muscular atrophy given the young age and the lack of upper motor neuron signs. This is a potentially ominous diagnosis in such a young patient, so I have, obviously, not discussed it with her at this point other than the fact that we need to pursue the Lyme disease workup. Should she have CSF criteria for Lyme disease, we will certainly treat her for it. If that is negative, then I think she will certainly deserve referral to a specialist for nerve and muscle biopsy and consideration of empiric treatment for Lyme disease. By her report, she has had symptoms for over seven years suggesting it is an indolent process which is more favorable but still quite concerning. I will keep you informed of further workup and follow-up and have discussed the workup at length with the patient and her mother although, as mentioned, I have not discussed specifically motor neuron disease at this point. Thank you for the opportunity to consult on this patient.

Sincerely,